Dr. Vishnu Shankar Sinha
Head, P G Department of Botany,
Tata College, Chaibasa (JH)

The Human Genome Project

The Human Genome Project (HGP) began in 1990 under supervision of **J. D. Watson** and subsequently by **Francis collions**, completed in 26th June, 2000. In human genome project there are six countries *i.e.,* USA,UK, France, Germany, Japan and China and 16 research laboratories with 1,100 scientists of life sciences and computer sciences are involved. The human genome project (HGP) was a international, collaborative research effort to map and sequence the entire human genome. HGP started with male chromosomes, because only males carry two different bundles of DNA *i.e.* 22A + Y, 22A + X.

Our body contains 100 million cells of over 260 different kinds *i.e.* brain cell, kidney cell, liver cell, lung cell etc. Nucleus of each cell contains 23 different chromosomes containing packed DNA excluding additional mitochondrial DNA.

In HGP, the most commonly used DNA markers or probes are restriction fragment length polymorphism (RFLP), variable number tandems repeats (VNTRs) or minisatellites and short tandem repeats (STRs) or microsatellites. Fluorescence in situ hybridization (FISH) is used to localize a gene on chromosome.

Human Genome contains about 3.2 giga bases or 3.2 billion base pairs but, only 1.1 - 1.5% of the genome is known to include the protein coding sequence (exons) of genes. Interestingly a human has perhaps 40,000 genes (arranged linearly along the chromosome) and we are 99.9% identical. Only one among 1000 chemical letters (A, C, G, T) of the genome of a person differ from that of another one.

TABLE : GENES PRESENT ON CHROMOSOMES

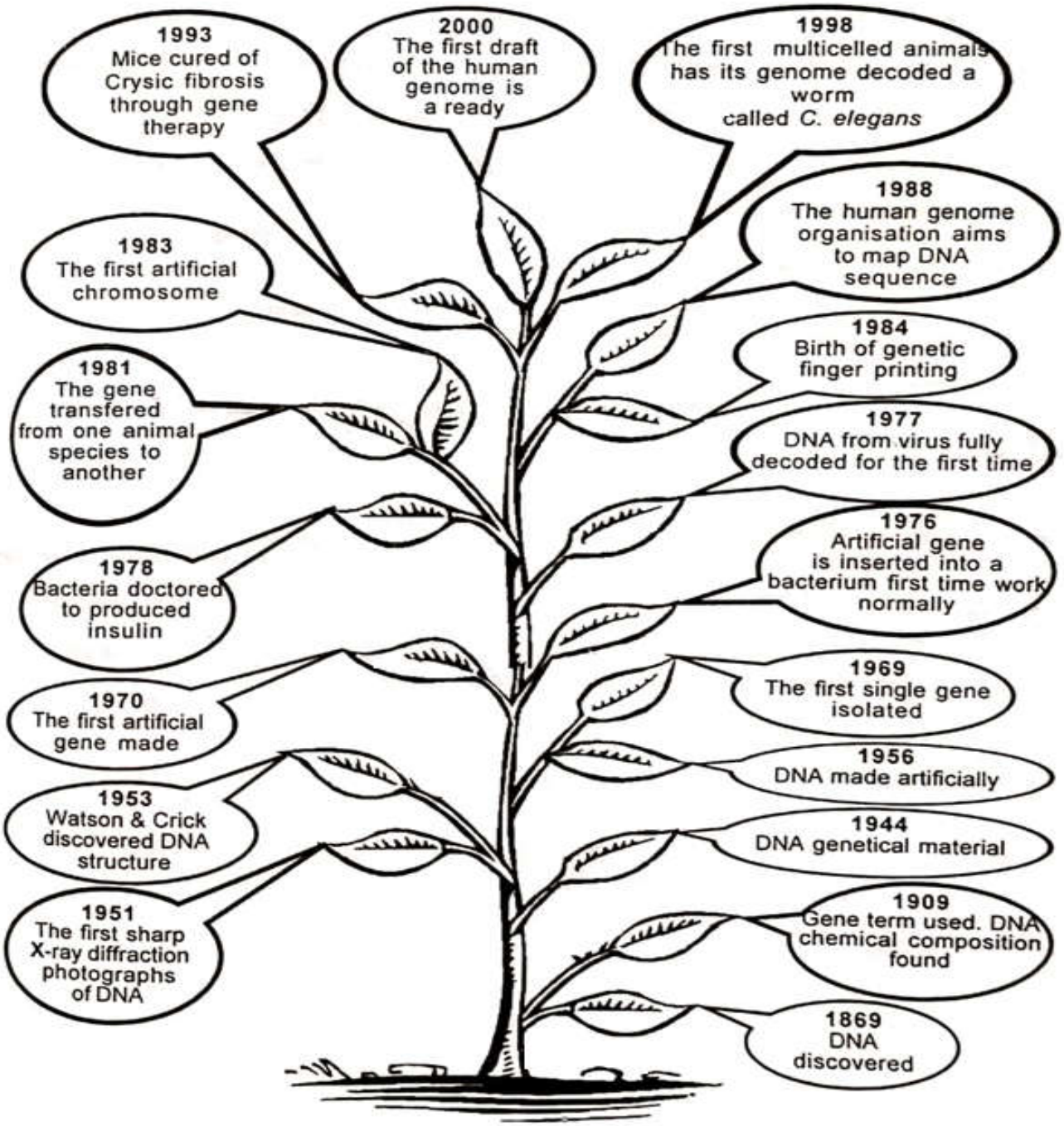
Chromosome Number	Gene Present	Function
1	**GBA**	Gaucher disease, absence of fat breaking enzyme, lead to jaundice and anemia.
	HPCLI	Prostate cancer.
	GLCIA	Glaucoma.
2	**EHM II**	Essential trimor (neurological disorder).
	MSH II	Colon cancer.
	CREB	Memory (Mice cannot learn without this gene).
	PAX III	Deafness and mismatched eye colour.
3	**VHL**	(Von Happil Lindu) Abnormal growth of blood vessel may develop in retina, certain area of brain, spinal cord, in the adrenal gland.
	SCLSI	Lung cancer.
	MLHI	Colon cancer (1, 60,400 American die in 1997).
4	**HD**	Huntington disease, degenerating brain, lead to dementia.
	EVC	Malformation of wrist bone, partial harelip and prenatal teeth eruption.
5	**SRD 51A**	Lead to baldness and acne.
	DTD	Malformations in joints.
6	**SCAI**	Loss of muscular co-ordination and spasticity.
	HLA	Responsible for organ transplantation from donor to recipient.
	IDDM I	Diabetes increases the risk of heart disease and kidney failure.

7	**GCK** **ELN** **OB** **GFTR**	Diabetes. Physical and mental development disorder (Williams's syndrome). Obesity. Cystic fibrosis.
8	**WRN** **MVC**	Premature aging occurring during adolescence (Burkitt lymphoma). Werner Syndrome, a rare form of cancer.
9	**CDKN II** **ABC I** **ABL** **TSC I**	Malignant melanoma. Tangler disease. Chronic myeloid leukemia. Tuberous sclerosis.
10	**PAHX** **GAT**	Failure of muscle co-ordination. Gyrate atrophy, an error of metabolism, loss of vision.
11	**LQT I** **IDDM II** **MEN I**	Long QT syndrome, can cause sudden cardiac death in young adults. Diabetes. Hyperactivity of endocrine gland.
12	**PAH**	Phenylketonuria
13	**BRCA-II** **RBI** **ATP7B**	Breast cancer. Retinoblastoma (eye tumor in children). Wilson disease, toxic copper accumulation (liver and neurological disease).
14	**PSI (AD3)**	Twice as common to women (memory loss).
15	**UBE 3A** **FBN I**	Angel man syndrome (mental retardation). Marfan syndrome (connective tissue disorder).
16	**FMF** **PKD I**	Familial Mediterranean fever. Polycystic kidney disease.
17	**P^{53}** **BRCA I**	Tumor-Suppressor protein. Breast cancer.
18	**NPCI** **DPC 4**	Nervous system impairment, common in children. Pancreatic cancer.
19	**APDF** **DM**	Atherosclerosis. Myotonic dystrophy (Mental deficiency, hair loss, cataracts).
20	**ADA I**	Severe combined immuno deficiency (no immunity to virus).
21	**SOD I** **APS I**	Amyotrophic lateral sclerosis. Autoimmune poly glandular syndrome.
22	**BCR**	Chronic myeloid leukemia (results when bone marrow replaced by Malignant leukemic cell).
23 **X Chr**	**DMD** **ATP7A** **(24,000 kbp)** **FMR I**	Duchene muscular dystrophy. Menkes syndrome (Severe cerebral). Fragile × Syndrome, mental retardation common in male.
23 **Y**	**SRY** **TDF (14 bp)**	Testis determining factor.

MAJOR FINDINGS OF THE HUMAN GENOME PROJECT:s

1. The human genome has approx 30,000–40,000 protein coding genes.
2. The human genome contains about 31664.7 million nucleotide bases.
3. Maximum number of genes has been reported in chromosome no 1 (8,000) and fewest no has been reported in the Y chromosome.
4. Only 1.1–1.5% of human genome consists of exons and about 98.5 – 98.9 % genome is noncoding (Introns).

5. Two different chromosomes of human genome differ widely with respect to number of genes per Mb, CpG islands, GC content, number of transposable element, recombination rate etc.
6. Human genome does more work than those of simpler organisms (Like fruit fly or round worm).
7. The human proteome is more complex than invertebrate's proteome.
8. Repeat sequences constitute more than 50% of the genome.
9. About 98% human DNA is identical to that of chimpanzees.
10. About 200 genes are close to that found in bacteria.



SIGNIFICANCE:

1. Production of molecular medicine.
2. Asses health damage and risk caused by explosive mutagenic chemicals and cancer causing toxin.
3. In Bioarchaeology, Anthropology, evolution and human migration.
4. DNA identification.
5. Implications for proteomics, Biotechnology and Bioinformatics.
6. Improve diagnostic testing: For disease susceptibility genes and for genes directly involved in the causation of specific diseases.
7. Benefitted the advancement of forensic science.

OBJECTIVES

1. Tumor suppressor protein is synthesised by the gene P^{53}, which is present on chromosome number
   (a) 6 (b) 7 (c) 13 (d) 17
2. How much part of human genome consist of exons (the protein coding region)
   (a) 10–15% (b) 3–5% (c) 1.1–1.5% (d) 0.1–5%
3. MHC, synthesised in response to the organ transplantation by the action of which gene
   (a) P^{53} (b) MLHI (c) HLA (d) CDKN-II
4. Human genome project helps in implication of
   (a) Proteomics (b) Biotechnology
   (c) Bio-inforamtics (d) All of these
5. Human genome project can be useful in improving human race by
   (a) Genetic councelling
   (b) Improving diagnostic testing for genes responsible for specific disease.
   (c) Production of moleculor medicine.
   (d) More than one
6. How many protein are coded by human genome ?
   (a) 20,000–25,000 (b) 30,000–40,000
   (c) 40,000–80,000 (d) 8,000–10,000
7. Till now the smallest gene of human being is
   (a) ATP7A (b) HLA (c) TDF (d) GBA
8. Diabetes increases the risk of heart and kidney failure due to IDDMI lie on
   (a) Chromosome no.6 (b) Chromosome no.9
   (c) Chromosome no.12 (d) Chromosome no.15
9. Accumulation of toxic copper lead to liver and neurological disease called Wilson disease due towhichgene
   (a) FMF (b) FBNI (c) ATP7B (d) BRCAII
10. TDF gene consist of
    (a) 14 bp (b) 24 bp (c) 34 bp (d) 44 bp
11. Breast cancer is caused by BRCA-I lie on chromosome number
    (a) 17 (b) 7 (c) 11 (d) 22
12. Inter-racial and inter-religion marriage lead to
    (a) Decrease gene pool (b) Increase gene pool
    (c) Constant gene pool (d) All of the above.